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CUTANEOUS PIGMENTATION AS AN INCOMPLETE FORM OF RECKLINGHAUSEN'S DISEASE, WITH REMARKS ON THE CLASSIFICATION OF INCOMPLETE AND ANOMALOUS FORMS OF RECKLINGHAUSEN'S DISEASE.

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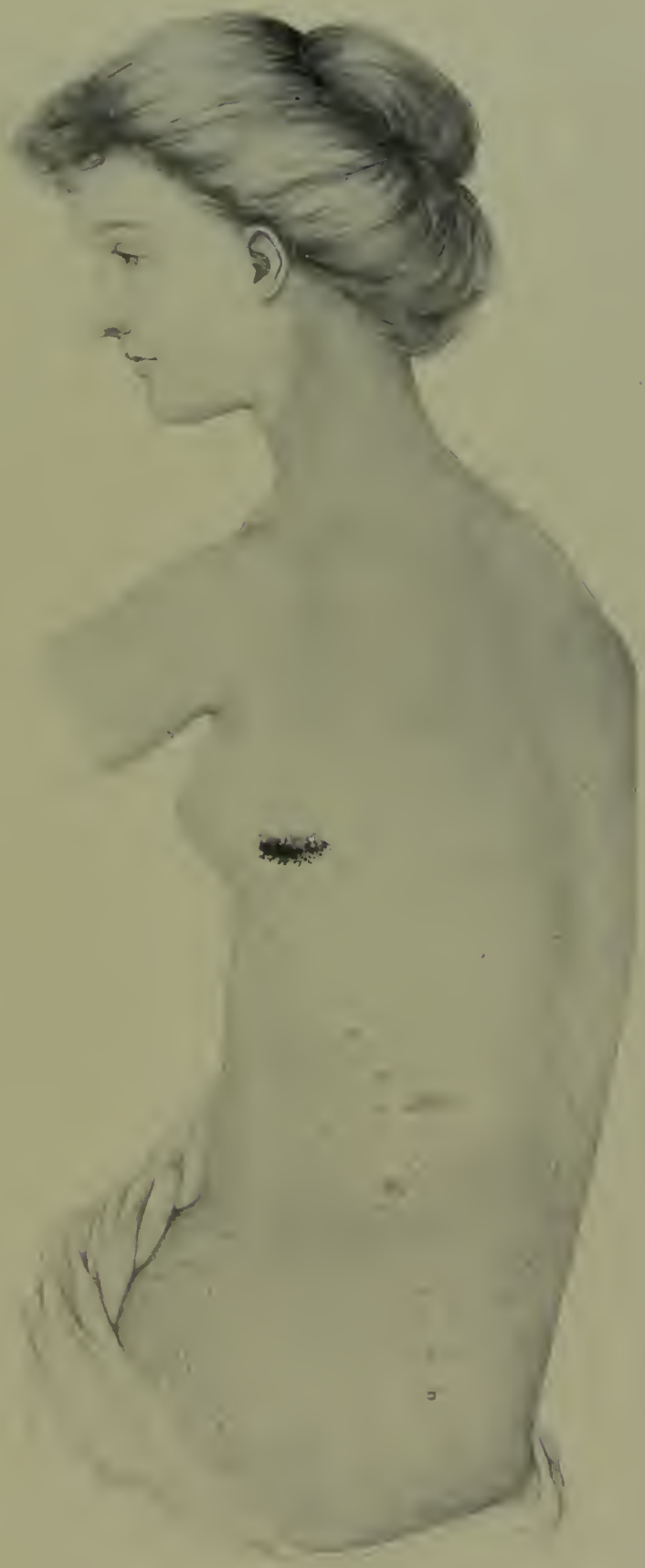
The patient, a fairly well-developed girl, was aged 14 years when I showed her case at the Dermatological Society of London, rather more than three and a half years ago, in May, 1905. The pigmentation affected the trunk and neck chiefly, and the extremities to a much lesser degree, the face being almost free. There was no evidence of urticaria, factitious urticaria, purpura, or any kind of erythema in connection with the condition. There were no areas of leucodermia. The patient (see figure, taken from a drawing by Miss Mabel Green in November, 1905), presented, roughly speaking, three different kinds of cutaneous pigmentation, namely: (1) Diffuse brownish patches, especially a large sheet of pigmentation over the upper part of the back and neck, which had a sharply defined upper border, but was ill-defined below, merging gradually into the ordinary skin; (2) brown spots and small patches plentifully scattered over the trunk, the paler ones being apparently those most recently developed; (3) a small group of very dark, almost black, spots on the left side of the thorax, resembling a group of "pigment nævi," but not raised above the general level of the skin. These last-mentioned blackish spots had been first noticed about three years previously. The earliest pigmentation noticed by the mother was at the back of the neck, when the patient was only about eighteen months old. Since that time the rest of the pigmentation had gradually developed, and fresh, relatively faint spots and patches had recently appeared on the extremities.

The patient's general nutrition was fairly good, and menstruation had lately commenced. Though she looked rather pale, examination of the blood showed nothing of pathological significance. The systolic brachial blood-pressure was found to be about 130 mm. mercury. Examination of the thoracic and abdominal viscera and of the urine showed nothing abnormal. Cutaneous sensation, knee-jerks and plantar reflexes were natural. The mental development and general intelligence were up to the average. There was no pigmentation in the mucous membrane of the mouth.

The only noteworthy point in the past history of the patient was that she had always been liable to headache and bilious vomiting. But there was no tendency to fainting, such as to suggest Addison's disease. The attacks of vomiting seemed rather to have been induced by certain articles of food, such as rich, fatty things, and, at one time, raw apples. Moreover, the patient's mother, as a child and young woman, had been subject to so-called "bilious attacks," and one or two other members of the family had been inclined to similar troubles.

Apparently no abnormal pigmentation had been observed in the family, except in the patient herself. In her case, from the diagnostic point of view, it was to be noted that the pigmentation affected chiefly the covered parts of the body, and that there was no special liability to freckling from exposure to the sun. This was the reverse of what one would expect in Kaposi's "Xerodermia pigmentosa." Moreover, in the present case, the variety in the colour and form of the areas of pigmentation and their situation (mainly on the trunk) made it seem most probable that the case was an incomplete form ("forme fruste") of Recklinghausen's disease—in fact, that it was a case of neuro-fibromatosis, with typical but excessive cutaneous pigmentation, though, as yet, practically without any (superficial) tumours. There was only a single, small, flaccid, molluscous tumour to be found. This was situated in the skin on the lower part of the back, and had first been observed about three years previously. Probably no other satisfactory explanation for the pigmentation could be offered except that given, namely, that it represented an early stage or incomplete form of Recklinghausen's disease—an explanation which was first suggested in a conversation by Dr. A. Whitfield.

When I last saw the patient, in July, 1908, the previous diagnosis



TO ILLUSTRATE DR. WEBER'S CASE OF INCOMPLETE RECKLINGHAUSEN'S DISEASE, IN NOVEMBER, 1905.

was confirmed by the formation of several other molluscous-like little tumours, which were developing in various parts of the skin. The pigmentation as a whole had become darker and new brownish spots had appeared. The skin of the upper and lower extremities was still relatively free, though there were some rather faint pale-brown patches on the arms and legs, larger than the ordinary macules on the trunk. The face was scarcely, if at all, affected, and there was no pigmentation of the mucous membrane of the mouth. No factitious urticaria could be obtained either over the normal or pigmented areas of skin. The systolic brachial blood-pressure was 130 mm. mercury, as before. The folds of the axillæ, elbows and knees were not specially pigmented, as they probably would have been in a case of Addison's disease. Dr. Graham Little, who kindly looked at the case with me, thought that the ulnar nerves at the elbows felt slightly enlarged, but I could not satisfy myself of this.

Cases of Recklinghausen's disease or neuro-fibromatosis undoubtedly occur, in which decided pigmentation of the skin is developed long before neuro-fibromata of nerve-trunks or molluscous tumours of the skin are observed. Thus, at the Medical Society of London, on November 9th, 1908, Dr. A. S. MacNalty showed a woman under Dr. Rose Bradford's care suffering from neuro-fibromatosis, in whom pigmentation had long been noticed before any neuro-fibromata were observed. A brother of this woman was supposed to show cutaneous pigmentation without any other evidence of disease. At a meeting of the Dermatological Section of the Royal Society of Medicine on November 19th, 1908, Dr. Graham Little showed a case of Recklinghausen's disease in a boy, aged 12 years, in whom, according to his mother's account, the cutaneous pigmentation preceded the development of the tumours by six years or more. In fact, some of the pigmentation seemed to date from birth. Undoubtedly, therefore, one has to admit the existence of an incomplete form, or early stage, of Recklinghausen's disease, characterised by scattered pigmented cutaneous spots and patches, which might range in size from minute macules, one or two millimetres in diameter, to large "sheets" extending from shoulder to shoulder, and might vary in the depth of the pigmentation from faint brown to almost black.

Though, strictly speaking, the term "Recklinghausen's disease" should be confined to cases showing (1) obvious neuro-fibromata in

connection with nerve-trunks, (2) molluscous tumours (mollusca fibrosa) of the skin, and (3) cutaneous pigmentation, yet incomplete or anomalous forms certainly occur in which one or even two of this triad of morbid features may be wanting. Moreover, there are likewise several cases on record in which multiple neuro-fibromata of nerve-trunks, or cutaneous neuro-fibromata (molluscous tumours), with or without cutaneous pigmentation, have been complicated by the co-existence of bony or epidermic (papillomatous) growth.

The following is a short scheme of classification for these anomalous or incomplete forms of Recklinghausen's disease :

(1) Cases of plexiform neuroma *unaccompanied by multiple molluscous tumours of the skin*, with or without cutaneous pigmentation. The swellings due to plexiform neuroma (Elephantiasis nervorum), which, perhaps, occur more frequently on the face and head than elsewhere, seem, on palpating them, as if they contained intertwined rootlets or tendrils ; and this is the reason why in Germany plexiform neuroma has been termed "Rankenneurom." Plexiform neuroma may show itself in the form of a very rare kind of macroglossia, as has been proved by the cases of Abbott and Shattock (1903) and Spencer and Shattock (1907),* a kind of macroglossia named "Macroglossia neuro-fibromatosa" by these authors.

(2) Cases of multiple molluscous tumours of the skin unaccompanied by any obvious neuro-fibromatosis of nerve trunks, with or without decided cutaneous pigmentation.

(3) Cases of pigmentation of the skin not (at least not yet) accompanied by obvious neuro-fibromata of nerve-trunks or cutaneous neuro-fibromata (molluscous tumours). This is the form of incomplete Recklinghausen's disease with which I have already dealt. It was not specially recognised, or given a special place, in Alexis Thomson's classical monograph, *On Neuroma and Neuro-fibromatosis*, published at Edinburgh in 1900.

(4) Anomalous cases of neuro-fibromatosis, complicated by the co-existence of bony or epidermic (papillomatous) changes. Benaky† described an example of this class under the heading "general neuro-fibromatosis, with molluscum pendulum of the right side of the face."

* Abbott and Shattock, *Path. Soc. Trans. Lond.*, vol. liv, p. 231 ; Spencer and Shattock, *Proc. Roy. Soc. Med.*, Pathological Section, vol. i, p. 8.

† *Ann. de Derm. et de Syph.*, November, 1904, p. 977.

In his case there were deformities of the cranium, vertebræ and tibia. In February, 1906, Mr. Ludford Cooper brought forward a case at the Ophthalmological Society, which he described as neuromatous elephantiasis, in a girl, aged 11 years. In Cooper's case the outer and lower portions of the frontal bone and the squamous portion of the temporal bone were much more prominent on the affected than on the other side. In 1901 Sir Jonathan Hutchinson* described and figured the case of a woman with multiple molluscous tumours of the skin, whose right cheek was bulged by bony overgrowth; the right frontal bone was likewise thickened, and the right eye was pushed forward as if by bony growth behind it. At the Society for the Study of Disease in Children, in April, 1907, Dr. Fletcher Beach† showed a case, described as one of Recklinghausen's disease, in a boy aged 5 years and 10 months, who apparently also had bony thickening in the right temporal region. The most extreme example of this class was doubtless the famous "elephant man," whom many must have seen when he was at the London Hospital. Sir Frederick Treves,‡ in his description of the "elephant man," mentioned that the deformities of the osseous system were limited to the skull, right upper extremity, and feet. "The proportions of the head were enormously increased, and its general outline was that of a hydrocephalic skull." There was exuberant papillomatous growth of some parts of his skin.

* *Polyclinic*, London, July, 1901, p. 12.

† *Reports of the Society for the Study of Disease in Children*, London, 1907, vol. vii, p. 167.

‡ *Path. Soc. Trans. Lond.*, 1885, vol. xxxvi, p. 494.